McGRAW-HILL PUBLICATIONS IN THE BOTANICAL SCIENCES

EDMUND W. SINNOTT, *Consulting Editor*

# *Principles of*
# GENETICS

PRINCIPLES OF GENETICS



VII

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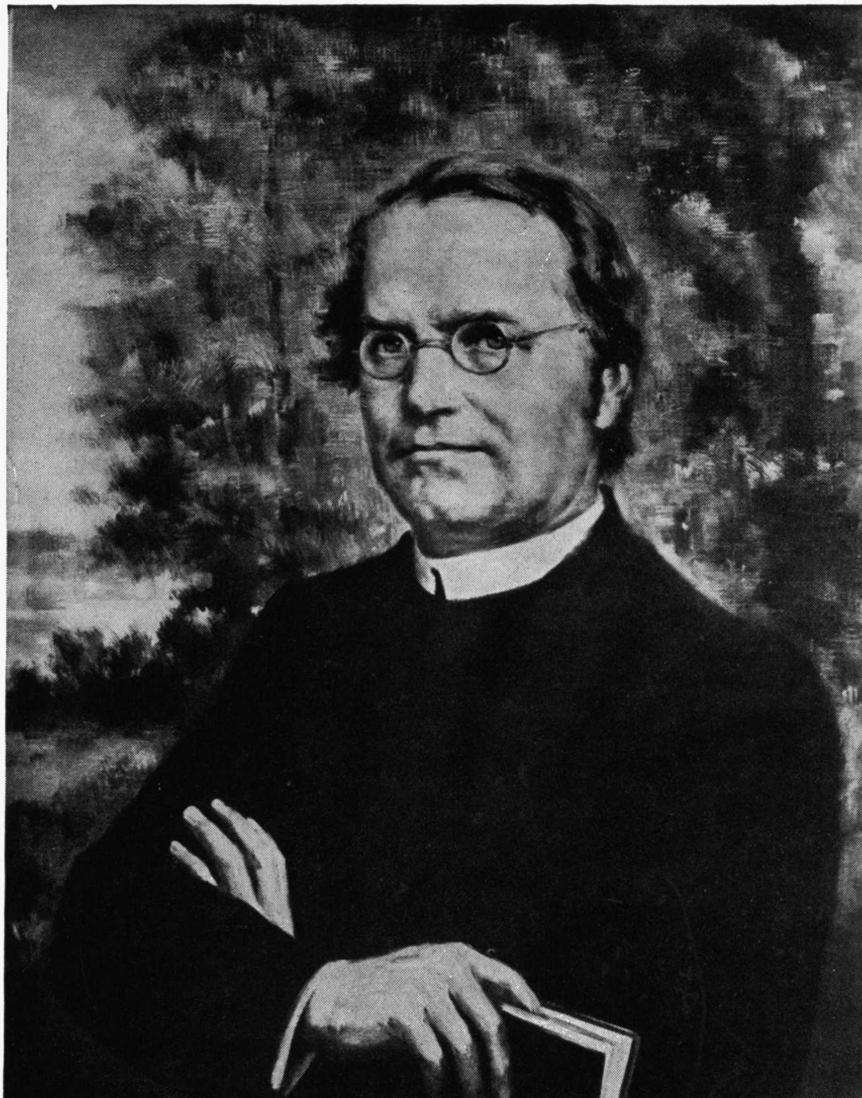

The founder of genetics, Gregor Johann Mendel, 1822–1884, from the painting by Flatter. (*Courtesy of Dr. Hugo Iltis.*)

## FOREWORD

There is a common feeling that a textbook is a full and final exposition of the subject which it treats, and that by virtue of "knowing the book" one acquires all the knowledge of the subject which it is necessary to have. Such beliefs have little to justify them. No text is or can be complete or final; nor, if it were, would an understanding of the subject be gained by committing the whole book to memory. Knowledge is not acquired in this way, but grows in the minds of those who discover for themselves new facts and relationships.

The principles of genetics have developed out of the arduous study of scores of investigators, and understanding of principles can best be gained by the student through a process which is somewhat similar to that employed in their original discovery. This process begins with, and is continually stimulated by, curiosity as to the methods and the mechanism of inheritance; it proceeds by the collection and study of facts, and by a critical discrimination between those which are true and relevant and those which are untrue or irrelevant; and finally it involves a considerable practice of the reasoning faculty by which deductions are made, and applied or tested on many similar cases. It is only in this way that the process of inheritance can be *understood.* The learning of facts alone cannot accomplish this.

As an aid to such a comprehension of the science of genetics, this book includes problems of two types, which form an integral part of the subject matter. These are designed to stimulate curiosity, to provide opportunity for practicing and extending the methods and applying the theories outlined in the text, and to point the way to other related facts not specifically treated in this book. They are not designed as memory tests, although the continual use of facts in solving problems is at once the best method of committing these facts to memory as well as of understanding them.

One of these aids consists of questions for thought and discussion. Answers to these are not to be found in the text itself, but may be reached by a process of reasoning for which only the premises are given. Familiarity with the subject matter of the text will provide the raw material, while the synthesis resulting in a correct answer or intelligent discussion must take place in the student's mind.

Other problems are designed to provide more extended practice in reasoning from principles. Nearly all of this type require some computation and may be most profitably studied as laboratory exercises under the guidance of an instructor. It is desirable to use labor-saving or "short-cut" methods (such as the checkerboard method described on page 63) wherever possible, in order that the mechanical work involved in calculation may not be regarded as the chief benefit to be derived from the problems. Sufficient information for solving all of them is contained in the text or in the supplementary notes in the problems.

The references cited will aid the student in examining the original publications from which our knowledge is derived. Of these Mendel's paper is still the most important and can be read with interest by all students. Citations of current literature are not intended to be complete; they should, however, indicate to the student that the subject as a whole is not contained in the text but is growing by the continual accretion of reports of experiments, all of which do not yield results in entire consonance with the few points of view which it is possible to present in a brief textbook. Some of the references will lead to new material not mentioned in the text, which must be reconciled with the fundamental principles of genetics, while others may serve to make connections between the student's knowledge of genetics and his experience in other directions.

# PREFACE

The fourth edition of this textbook was called for not only by the many additions to our knowledge of genetics which the past ten years have brought but also by the need for correcting and strengthening those sections of the third edition which both we and many of our colleagues had recognized as inadequate.

Although the entire book has been rewritten, we have tried to preserve its essential character as a general introduction to principles and the actual problems of genetics. The chief changes will be found in the chapters dealing with the physical basis of heredity in the chromosomes (Chapters VII through X). We have described the chromosome mechanism not for its own sake but as a genetical system, the transmission apparatus of heredity, and have described the departures from the normal structure and functioning of this system (chromosome aberrations) in some detail because from such abnormalities proofs of the actual arrangements of genes in chromosomes, the cytological maps, can be obtained.

Most of the new material added concerns the fields of special interest to the two authors chiefly responsible for the revision, namely, population genetics and speciation on the one hand, and on the other, the mechanisms of genic effects on development. It happens that the chief recent expansions in our knowledge of genetics have occurred in these fields. We have devoted a good deal of space to the use of fungi such as Neurospora, and microorganisms such as bacteria, protozoa, and viruses, which has opened up possibilities for understanding the mode of action of genes. The analysis of the breeding structure of natural populations in both animals and plants and of human populations has required the insertion of three chapters (XII, XIII, and XIV) dealing with the general principles of population genetics based on the Hardy-Weinberg equilibrium, on race formation, and on speciation.

Throughout the book the use of material and examples from human heredity has increased considerably over that in previous editions. Instead of separating this from the other materials of genetics we have treated it as a part of a consistent whole, for heredity in man differs in no essential way from heredity in other animals and plants.

The problems have been revised and many new ones introduced, especially in the chapters on chromosomal aberrations and population genetics.

As an appendix we have included a translation of the original report of Mendel (1865), with which modern genetics began. We believe all students of genetics should be familiar with this paper which still forms the best introduction to the basic evidence and the principles derived from it. The translation was made by the Royal Horticultural Society of London, and we are grateful to the council of the society for permission to reproduce it.

We are indebted to many colleagues for their courtesy in supplying new illustrations as acknowledged in the captions, and particularly to Professor M. M. Rhoades for criticism and for new material prepared for this edition, including the photomicrographs of meiosis in maize and the linkage maps of maize. We acknowledge with special gratitude the aid and patience, in connection with the preparation of the manuscript, of Natalie Sivertsev Dobzhansky, Louise Porter Dunn, and Pauline Goldman.

THE AUTHORS

*April*, 1950

# CONTENTS

# CHAPTER I

## GENETICS, THE SCIENCE OF HEREDITY AND VARIATION

Between those things which are alive and those which are lifeless there exists a gap which science has not yet bridged. Living beings which are so various as to include man and the higher and lower animals, the great plant kingdom, the microorganisms, and the smallest forms of all, the viruses, all share certain common properties which distinguish them from lifeless matter. One of the most important of these properties is self-reproduction. The organized unit in which living matter always occurs—that is, the living individual—must always arise from some preexisting living individual and never from lifeless matter itself.

The classical work of Spallanzani and of Pasteur gave the deathblow to the old belief in the "spontaneous generation" of living things out of dead material and proved that even among the most minute organisms the spark of life can be kindled only by life itself. Recent studies on viruses which multiply in the living cells of plants and animals, seem to have narrowed somewhat the gap between the living and the lifeless, since, under the electron microscope, aggregates of some viruses appear as crystallike bodies. Yet, so far as we know now, individual virus particles arise only from living virus. Every organism now living is therefore to be looked upon as the latest member of a long and uninterrupted succession of living beings, extending back, generation after generation, to the dawn of life. This is the essential teaching of the theory of evolution. The actual origin of life itself is lost in the mists of antiquity, but the pageant of the evolutionary history of living things, which unfolds itself in the fossil record of ancient times, makes it clear beyond any reasonable doubt that the animals and plants of today are direct lineal descendants of earlier and more primitive types. Continuity is of the essence of life.

**Reproduction.** Since individual living things grow old and die, however, this continuity must be maintained by the transmission of life from one individual to a succession of new ones, its offspring. This process is known as *reproduction* and may take place in various ways.

In the simplest methods, commonly called *asexual*, or *vegetative*, reproduction, the body of the parent becomes divided into two or more parts, each of which grows into a new individual. With animals this method is uncommon except in the simplest types, but among plants the fact that a small portion of the body, when removed and placed under favorable conditions, will often restore the missing parts and establish itself as a new individual makes multiplication of this type easy and effective both in nature and through the various arts of plant propagation.

Far commoner and more important than this asexual, or vegetative, method is that called *sexual reproduction*. An essential feature here is that the new individual arises through the union of two sex cells, or *gametes* which form one cell, or *zygote*, from which develops the new individual. The successful consummation of this process is ensured by a great variety of structures and functions found throughout the animal and plant kingdoms. In the lowest plants and animals the gametes which unite may be so much alike that neither of them can be classed as female or male. In most organisms, there is a division of labor between relatively large, food-laden female cells, the *eggs*, or *ova*, and small and usually motile male cells, known as *sperms*, or *spermatozoa*. In the higher plants a series of complicated reproductive structures—the flower, fruit, and seed—have been evolved. The male gametes are here produced within *pollen grains* and the female gametes within the *ovules*. The fertilized egg develops into the embryo of the seed.

**Heredity.** The single gamete which a parent contributes to each of its offspring is usually too small to be visible to the unaided eye (Fig. 1). Yet this extremely narrow bridge is the only direct physical link between parents and offspring, and across it everything must pass which is *transmitted* from one generation to the next. Muller has estimated that all the spermatozoa from which the present population of the world arose (more than 2 billion individuals) would make no larger bulk than half of an ordinary aspirin tablet. The essential parts, that is, the chromosomes, of 2 billion eggs would occupy about the same space. This minute amount of substance has nevertheless determined, in cooperation with the factors of the environment, the kinds of human beings which inhabit the earth. Indeed, these tiny containers of reproductive substance, into which so much is packed and out of which so much emerges, are the most remarkable bits of matter in existence.

Regardless of whether an organism reproduces sexually or asexually, the bit of the parental body which gives rise to the new individual undergoes growth. The body of an adult man has a mass tens of billions of times greater than that of the fertilized egg from which it has developed. In order for growth to occur, much material has evidently to be taken from the environment and incorporated into the developing body. In the case of green plants, this material consists of water and mineral salts taken from the soil and of carbon dioxide and oxygen from the air, the energy of sunlight making the assimilation of these other materials possible. In living things other than green plants, growth takes place by the incorporation into the body of whatever organic and inorganic foods the organism requires.

The food taken in from the environment becomes a part of a living body

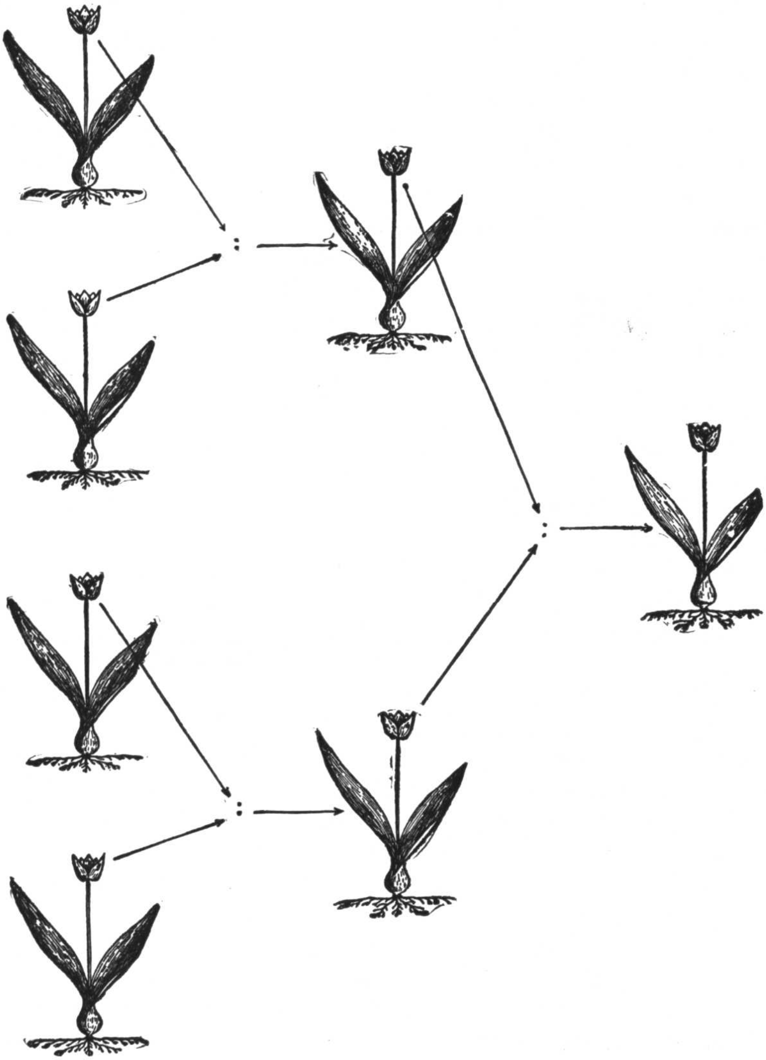

FIG. 1. The narrow hereditary bridge. The plant at the right receives from each of its parents only one minute sexual cell, a male gamete from one and a female gamete from the other. The parents, in turn, receive from each of the grandparents but one sexual cell. Thus the bridge which connects one generation with the next, and over which the entire inheritance must pass, is an exceedingly narrow one.

only through profound chemical and physical transformations wrought by the body upon the substances in the food. Moreover, and this is fundamental, every organism grows by transforming its food in a definite way, so that the outcome of the growth process is always a more or less faithful copy of the bodies of the parents and other ancestors of the developing individual. Thus, the parental organism reproduces itself in its offspring by causing it to organize, in the same definite way, the materials taken in from the environment; and this process of self-reproduction is the essence of *heredity.* It is because of heredity that individuals related by descent resemble each other.

**Variation.** Heredity does not necessarily mean that parents and offspring are completely identical. Probably no two human beings or two individuals of any other species are ever exactly alike. This is, first of all, because the environment of organisms is never the same in different places and at different times. No two plants which grow side by side in a meadow receive precisely the same amounts of light, water, and minerals; no two animals receive quite the same food at the same stage of development. Two individuals with the same heredity may become somewhat different when they come into contact with different conditions of food, temperature, light, humidity, and other external factors. Such differences among organisms of similar heredity are referred to as *environmental variations*, or *modifications.*

The second cause of unlikenesses is that there are many different kinds of hereditary constitution. Although each kind tends to reproduce itself faithfully from generation to generation, changes in heredity occasionally do occur. These changes, called *mutations*, are perpetuated in the offspring, because the altered heredity reproduces itself just as faithfully as the original condition did. It will be shown later that the hereditary substance is composed of discrete parts, called *genes*, and that different genes may undergo mutation independently. In the process of sexual reproduction, different genes become associated with each other in various combinations. *Recombination* is one of the commonest sources of hereditary variability. The numbers of these combinations may be so enormous that, in most sexually reproducing species including man, probably no two individuals (except identical twins, see page 20) have exactly the same heredities. The diversity which is produced by differences in heredity is described as hereditary variation, or *genotypic variation.* Environmental and hereditary variations occur in nature side by side, so that the differences observed between individuals are usually partly environmental and partly hereditary in origin.

**Evolution.** Because of its peculiar hereditary constitution, each organism from the lowest virus to the highest animal tends to transform the materials from the environment into its own likeness; and as reproduction continues and the descendants increase, the process exercises a "pressure" on the environment. This pressure tends to transform the materials available in the world into bodies of a particular kind of organism. The first living thing that appeared on earth started this trend toward converting nonliving into living matter.

The rates of reproduction differ widely in different organisms. Some bacteria divide once in about twenty minutes. The length of a generation in man is about a quarter of a century, and the number of offspring produced by a pair is small. A sequoia tree lives for three thousand years or more and produces numerous seeds. Nevertheless, any organism tends to multiply until it exhausts the available food supply, occupies all the accessible space, or is checked by enemies or parasites. Each species can use only certain substances for food and can inhabit only certain climates and soils. To be sure, some forms of life are more and others less narrowly specialized in food and habitat requirements. Thus, some parasitic protozoans live only in the blood stream of a single species of a mammal or a bird; on the other hand, the creosote bush, *Larrea divaricata*, grows apparently equally well below sea level or very high in the mountains. However that may be, the populations of any species of organism tend to expand until the expansion is checked by the inertia of the environment.

We have seen that the conservatism of heredity, which makes like beget like, is opposed by mutation, which produces new hereditary variants. Some of these variants may be able to use as food, substances which are not used or which are exploited less efficiently by other organisms. One type of variant may reproduce faster than others under certain climatic or soil conditions. If the new variant reproduces more efficiently, it will gradually supplant the ancestral form; or else both the original and the ancestral forms will exist, each in the sphere in which it is most competent. In either case, there will have appeared in the world a new form of life. On the other hand, a variant which reproduces less efficiently than the original form (and most variants that arise by mutation are less efficient) will be eliminated. The propagation and spread of the efficient and the nonperpetuation of the inefficient hereditary variants constitute what is known as *natural selection.*

Heredity modified by one mutation may be changed again and again by other mutations. Darwin in "The Origin of Species," published in 1859, developed a theory which, in our modern terminology, may be stated as follows: Natural selection, acting on a succession of hereditary variants produced by mutation, leads to a gradual change, or *evolution*, of organisms.

It may also be said that evolution is a necessary outcome of the interaction of two opposite forces: heredity, the conservative agent which makes